



PNP gene

purine nucleoside phosphorylase

Normal Function

The *PNP* gene provides instructions for making an enzyme called purine nucleoside phosphorylase. This enzyme is found throughout the body but is most active in specialized white blood cells called lymphocytes. These cells protect the body against potentially harmful invaders by making immune proteins called antibodies that tag foreign particles and germs for destruction or by directly attacking virus-infected cells. Lymphocytes are produced in specialized lymphoid tissues including the thymus and lymph nodes, and then released into the blood. The thymus is a gland located behind the breastbone; lymph nodes are found throughout the body. Lymphocytes in the blood and in lymphoid tissues make up the immune system.

Purine nucleoside phosphorylase is known as a housekeeping enzyme because it clears away waste molecules called deoxyinosine and deoxyguanosine, which are generated when DNA is broken down. Specifically, purine nucleoside phosphorylase converts deoxyinosine to another molecule called hypoxanthine, and converts deoxyguanosine to another molecule called guanine.

Health Conditions Related to Genetic Changes

purine nucleoside phosphorylase deficiency

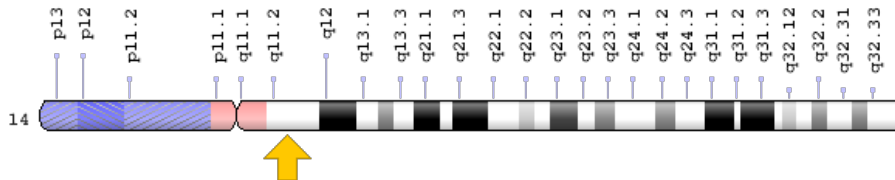
At least 24 *PNP* gene mutations have been identified in individuals with purine nucleoside phosphorylase deficiency. Most of these mutations change single protein building blocks (amino acids) in the purine nucleoside phosphorylase enzyme. The mutations reduce or eliminate the activity of purine nucleoside phosphorylase. The resulting excess of waste molecules and further reactions involving them lead to the buildup of a substance called deoxyguanosine triphosphate (dGTP) to levels that are toxic to lymphocytes.

Immature lymphocytes in the thymus are particularly vulnerable to a toxic buildup of dGTP, which damages them and triggers their self-destruction (apoptosis). The number of lymphocytes in other lymphoid tissues is also greatly reduced, resulting in the immune deficiency and vulnerability to severe infections characteristic of purine nucleoside phosphorylase deficiency.

Chromosomal Location

Cytogenetic Location: 14q11.2, which is the long (q) arm of chromosome 14 at position 11.2

Molecular Location: base pairs 20,469,379 to 20,478,006 on chromosome 14 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- inosine phosphorylase
- NP
- PNPH_HUMAN
- PRO1837
- PUNP
- purine-nucleoside:orthophosphate ribosyltransferase

Additional Information & Resources

Educational Resources

- Immunobiology (fifth edition, 2001): Defects in T-Cell Function Result in Severe Combined Immunodeficiencies
<https://www.ncbi.nlm.nih.gov/books/NBK27109/#A1509>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28purine+nucleoside+phosphorylase%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D>

OMIM

- PURINE NUCLEOSIDE PHOSPHORYLASE
<http://omim.org/entry/164050>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_PNP.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=PNP%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=7892
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/4860>
- UniProt
<http://www.uniprot.org/uniprot/P00491>

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